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学位論文の題名	Polo-like kinase 4 and stromal antigen 3 are not associated with recurrent pregnancy loss caused by embryonic aneuploidy （Polo-like kinase 4・Stromal antigen 3(STAG3)と絨毛染色体異常による不育症との関連性について） Human Genome Variation, 29;7:18, 2020
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Abstract

Objective: No genetic association with recurrent pregnancy loss (RPL) caused by embryonic aneuploidy has been found. Recent studies have indicated that the common genetic variant rs2305957, surrounding the PLK4 gene, contributes to mitotic-origin aneuploidy risk during human early embryo development. The decrease in meiosis-specific cohesin causes predivision of sister chromatids in the centromere and chromosome segregation errors. STAG3 is a component of cohesin and is a meiosis-specific gene.

Methods: Our case-control study included 184 patients with RPL whose previous products of conception (POC) exhibited aneuploidy and 190 fertile control women without a history of miscarriage. We performed a genetic association study to examine the genotype distribution at PLK4 (rs2305957) and STAG3 in patients with RPL caused by aneuploidy compared with controls. Regarding STAG3, SNPs with a minor allele frequency (MAF) threshold > 0.05 that were predicted to be binding sites of transcription factors and that showed significant associations in expression quantitative trait locus (e-QTL) analysis were selected.

Results: No significant differences in the MAF or distribution in any model of PLK4 (rs2305957) and 5 selected tag SNPs in STAG3 were found between the patients and controls.

Conclusions: A further genome-wide association study is needed since a combination of genetic risk alleles might be useful in predicting future age-dependent RPL caused by aneuploidy.